

## **DIAGNOSTIC METHODS FOR POMPE DISEASE AND OTHER LYSOSOMAL STORAGE DISEASES**

### **Abstract**

- 5            Provided are methods of screening subjects for lysosomal storage diseases, preferably glycogen storage diseases, using a tetrasaccharide as a biomarker. In a more preferred embodiment, subjects are screened for Pompe disease (*i.e.*, glycogen storage disease type II). Also provided are neonatal screening assays. The present invention further provides methods
- 10 of monitoring the clinical condition and efficacy of therapeutic treatment in affected subjects. Further provided are methods of measuring a tetrasaccharide biomarker by tandem mass spectrometry, preferably, as part of a neonatal screening assay for Pompe disease.